

## Author Index to Volume 50

(ASHG) = American Society of Human Genetics report; (BR) = book review; (CHG-PA) = Congress of Human Genetics presidential address; (CHG-PL) = Congress of Human Genetics plenary lecture; (E) = editorial; (L) = letter to the editor; (Obit) = obituary; (Op) = opinion

- Abel, L., 1308  
 Abeliovich, D., 1185  
 Adams, K. J., 1129  
 Adelsberger, P. A., 288, 544  
 Affara, N. A., 317  
 Akkari, P. A., 576  
 Akots, G., 607  
 Allamand, V., 634  
 Allsopp, C. E. M., 411  
 Alper, J. K., 465  
 Alper, J. S., 465, 476  
 Alperovitch, A., 645 (L)  
 Amini, S., 1252  
 Amos, J., 902  
 Andrew, S., 382  
 Anstey, N., 411  
 Antonarakis, S. E., 15, 288, 544, 607, 1038  
 Anvret, M., 278  
 Appleton, R. E., 508  
 Arahata, K., 896  
 Arnheim, N., 347  
 Arnos, K. S., 902  
 Artlich, A., 876 (L)  
 Asher, J. H., Jr., 902  
 Ashton, L. J., 787  
 Assmann, G., 1115  
 Atif, U., 590  
 Augarten, A., 222  
 Austin, M. J. F., 76  
 Azen, E. A., 842
- Baird, J., 607  
 Baldini, A., 826  
 Ballabio, A., 483  
 Ballard, S. G., 71  
 Bao, Y., 229  
 Barker, D. F., 42  
 Barnett, A. H., 1018  
 Bartels, C. F., 1086, 1104  
 Bartsch, G., 1318  
 Bartsocas, C., 544  
 Basaran, S., 288  
 Bashan, N., 222
- Bashir, R., 590  
 Basler, E., 483  
 Bassand, P., 1281  
 Bateman, J. B., 446 (L)  
 Baumiller, R. C., 239 (BR)  
 Beaty, T. H., 1038  
 Beaudet, A. L., 439 (L)  
 Beck, J. S., 567  
 Beckmann, J. S., 1211  
 Beckwith, J., 476  
 Beggs, A. H., 576  
 Beighton, P., 902  
 Ben-Yoseph, Y., 137  
 Bernhardt, B. A., 84  
 Bessis, J.-L., 364  
 Bétard, C., 92  
 Beutler, E., 394  
 Bhattacharya, S. S., 590, 876 (L)  
 Biancalana, V., 981  
 Biesecker, L., 438 (L)  
 Billings, P. R., 476  
 Binkert, F., 288, 544  
 Birch, P. H., 1129  
 Bird, A. C., 590  
 Bird, T. D., 42, 896  
 Black, D. M., 1235  
 Blakemore, K. J., 700  
 Blasi, F., 492  
 Bleeker-Wagemakers, E. M., 1195  
 Blonden, L. A. J., 317  
 Blumberg, P. C., 576  
 Bockel, B., 652 (L)  
 Bodis-Wollner, I., 443 (L)  
 Bodmer, W. F., 671  
 Bolund, L., 492  
 Boman, H., 15, 360  
 Bonaiti, C., 645 (L)  
 Bonaiti-Pellie, C., 1231  
 Bonilla, E., 934  
 Bonizzato, A., 1140 (L)  
 Børghlum, A. D., 492  
 Bouffard, O., 92  
 Bouhassira, E. E., 71
- Boundy, K. L., 576  
 Bowcock, A. M., 371  
 Bowden, D. W., 607  
 Bowles-Biesecker, B., 438 (L)  
 Bozon, D., 1178  
 Brackmann, H. H., 164  
 Bradley, C., 294  
 Bradwell, A. R., 1018  
 Brasseur, R., 208  
 Breakefield, X. O., 619  
 Brenner, D. A., 1203  
 Bressman, S. B., 619  
 Brewster, D., 411  
 Briard, M. L., 981  
 Brin, M. F., 619  
 Brock, D., 607  
 Brock, D. J. H., 749  
 Brockington, M., 629  
 Brooks, D. A., 787  
 Brown, J. A., 76  
 Brown, R. H., Jr., 896  
 Brunsmann, F., 1195  
 Bryke, C. R., 988  
 Buckle, V. J., 520  
 Budarf, M. L., 924  
 Budowle, B., 441 (L), 816  
 Bull, L., 1218  
 Bulman, D. E., 950  
 Bunce, M., 411  
 Burghes, A. H. M., 950  
 Burke, R. E., 619  
 Burlet, P., 892  
 Burn, J., 1340 (L)  
 Butler, I. J., 677  
 Bütler, R., 208  
 Byrne, E., 1337 (L)  
 Byskov, A., 492
- Camerino, G., 156  
 Cao, A., 422  
 Carestia, C., 781  
 Carmen, I. H., 245 (Op)  
 Carnevale, P., 1308  
 Carrasco, S., 1022

## Author Index to Volume 50

(ASHG) = American Society of Human Genetics report; (BR) = book review; (CHG-PA) = Congress of Human Genetics presidential address; (CHG-PL) = Congress of Human Genetics plenary lecture; (E) = editorial; (L) = letter to the editor; (Obit) = obituary; (Op) = opinion

- Abel, L., 1308  
 Abeliovich, D., 1185  
 Adams, K. J., 1129  
 Adelsberger, P. A., 288, 544  
 Affara, N. A., 317  
 Akkari, P. A., 576  
 Akots, G., 607  
 Allamand, V., 634  
 Allsopp, C. E. M., 411  
 Alper, J. K., 465  
 Alper, J. S., 465, 476  
 Alperovitch, A., 645 (L)  
 Amini, S., 1252  
 Amos, J., 902  
 Andrew, S., 382  
 Anstey, N., 411  
 Antonarakis, S. E., 15, 288, 544, 607, 1038  
 Anvret, M., 278  
 Appleton, R. E., 508  
 Arahata, K., 896  
 Arnheim, N., 347  
 Arnos, K. S., 902  
 Artlich, A., 876 (L)  
 Asher, J. H., Jr., 902  
 Ashton, L. J., 787  
 Assmann, G., 1115  
 Atif, U., 590  
 Augarten, A., 222  
 Austin, M. J. F., 76  
 Azen, E. A., 842  
 Baird, J., 607  
 Baldini, A., 826  
 Ballabio, A., 483  
 Ballard, S. G., 71  
 Bao, Y., 229  
 Barker, D. F., 42  
 Barnett, A. H., 1018  
 Bartels, C. F., 1086, 1104  
 Bartsch, G., 1318  
 Bartsocas, C., 544  
 Basaran, S., 288  
 Bashan, N., 222  
 Bashir, R., 590  
 Basler, E., 483  
 Bassand, P., 1281  
 Bateman, J. B., 446 (L)  
 Baumiller, R. C., 239 (BR)  
 Beaty, T. H., 1038  
 Beaudet, A. L., 439 (L)  
 Beck, J. S., 567  
 Beckmann, J. S., 1211  
 Beckwith, J., 476  
 Beggs, A. H., 576  
 Beighton, P., 902  
 Ben-Yoseph, Y., 137  
 Bernhardt, B. A., 84  
 Bessis, J.-L., 364  
 Bétard, C., 92  
 Beutler, E., 394  
 Bhattacharya, S. S., 590, 876 (L)  
 Biancalana, V., 981  
 Biesecker, L., 438 (L)  
 Billings, P. R., 476  
 Binkert, F., 288, 544  
 Birch, P. H., 1129  
 Bird, A. C., 590  
 Bird, T. D., 42, 896  
 Black, D. M., 1235  
 Blakemore, K. J., 700  
 Blasi, F., 492  
 Bleeker-Wagemakers, E. M., 1195  
 Blonden, L. A. J., 317  
 Blumberg, P. C., 576  
 Bockel, B., 652 (L)  
 Bodis-Wollner, I., 443 (L)  
 Bodmer, W. F., 671  
 Bolund, L., 492  
 Boman, H., 15, 360  
 Bonaiti, C., 645 (L)  
 Bonaiti-Pellie, C., 1231  
 Bonilla, E., 934  
 Bonizzato, A., 1140 (L)  
 Børghlum, A. D., 492  
 Bouffard, O., 92  
 Bouhassira, E. E., 71  
 Boundy, K. L., 576  
 Bowcock, A. M., 371  
 Bowden, D. W., 607  
 Bowles-Biesecker, B., 438 (L)  
 Bozon, D., 1178  
 Brackmann, H. H., 164  
 Bradley, C., 294  
 Bradwell, A. R., 1018  
 Brasseur, R., 208  
 Breakefield, X. O., 619  
 Brenner, D. A., 1203  
 Bressman, S. B., 619  
 Brewster, D., 411  
 Briard, M. L., 981  
 Brin, M. F., 619  
 Brock, D., 607  
 Brock, D. J. H., 749  
 Brockington, M., 629  
 Brooks, D. A., 787  
 Brown, J. A., 76  
 Brown, R. H., Jr., 896  
 Brunsmann, F., 1195  
 Bryke, C. R., 988  
 Buckle, V. J., 520  
 Budarf, M. L., 924  
 Budowle, B., 441 (L), 816  
 Bull, L., 1218  
 Bulman, D. E., 950  
 Bunce, M., 411  
 Burghes, A. H. M., 950  
 Burke, R. E., 619  
 Burlet, P., 892  
 Burn, J., 1340 (L)  
 Butler, I. J., 677  
 Bütler, R., 208  
 Byrne, E., 1337 (L)  
 Byskov, A., 492  
 Camerino, G., 156  
 Cao, A., 422  
 Carestia, C., 781  
 Carmen, I. H., 245 (Op)  
 Carnevale, P., 1308  
 Carrasco, S., 1022

- Casals, T., 404, 1140 (L)  
 Caskey, C. T., 1143 (BR)  
 Cassani, G., 492  
 Cederbaum, S. D., 1281  
 Chakraborty, R., 145  
 Chakravarti, A., 544, 1031  
 Chamberland, A., 92  
 Chan, L., 1275  
 Chance, P. F., 42  
 Chartier-Harlin, M.-C., 648 (L)  
 Chen, T.-H., 137  
 Chen, Y.-T., 229  
 Cheng, T.-Y., 237 (L)  
 Chessa, L., 1343 (L)  
 Childs, B., 240 (BR)  
 Chillón, M., 1140 (L)  
 Choo, K. H. A., 706, 717  
 Christensen, S. R., 1203  
 Christodoulou, J., 852  
 Cikuli, M., 875 (L)  
 Clarke, J. T. R., 852  
 Claussen, U., 1031  
 Cleaver, J. E., 677  
 Clements, P. R., 787  
 Clerget-Darpoux, F., 645 (L), 892, 1231  
 Coetzee, G. A., 427  
 Cogen, P. H., 584  
 Cohen, D., 1211  
 Cohen, M. M., 544  
 Collin, C., 382  
 Collins, F., 438 (L)  
 Collins, J. M., 76  
 Coloma, A., 434  
 Conneally, P. M., 528, 536  
 Connor, L., 646 (L)  
 Coovert, D. D., 950  
 Coresh, J., 1038  
 Corey, L. A., 76  
 Corey, M., 1178  
 Cormier, V., 364  
 Costa, P., 1027  
 Costa, P. P., 1027  
 Cot, M., 1308  
 Cotton, R., 875 (L)  
 Cousineau, A., 567  
 Cox, D. R., 1218  
 Crall, M., 190  
 Crawford, M., 875 (L)  
 Cremers, F. P. M., 1195  
 Cubey, R. B., 1340 (L)  
 Cupples, L. A., 528, 646 (L), 653 (L)  
 Curristin, S. M., 1185  
 Cutting, G. R., 1185  
 Dailey, H. A., 1203  
 Dale, S., 717  
 Dallapiccola, B., 875 (L)  
 Daneshvar, L., 584  
 Daniels, R. J., 520  
 Danø, K., 492  
 David, A., 981  
 Davies, K. E., 520, 887 (E)  
 Davignon, J., 92  
 Davis, A., 190  
 Davis, M. B., 629  
 De Angioletti, M., 781  
 de Bonis, C., 781  
 de Cuevas, M., 476  
 DeGennaro, L. J., 278  
 de Leon, D., 619  
 Denniston, C., 1145 (BR)  
 De Rosa, L., 781  
 Desnick, R. J., 795  
 Deufel, T., 1151  
 DeVries, A., 1151  
 Didier, J. M., 1203  
 Diehl, S. R., 902, 1259  
 DiMauro, S., 934  
 Ding, J.-H., 229  
 Distèche, C. M., 294  
 Dobyns, W. B., 182  
 Dodson, A. E., 1281  
 Dognini, M., 1140 (L)  
 Donis-Keller, H., 1267  
 Donnelly, A., 968  
 do Rosario Almeida, M., 1027  
 Dozy, A., 422  
 Driscoll, D. A., 924  
 Duclos, F., 559  
 Duffy, B., 1348 (L)  
 Duke-Woodside, M. E., 677  
 Dunning, A. M., 208  
 Durie, P., 1178  
 Duyk, G., 584  
 Earle, E., 706, 717  
 Eberle, J., 1318  
 Edwards, M. S. B., 584  
 Ehnholm, C., 816  
 Eiken, H. G., 360  
 Elias, S., 6  
 Ellis, J. M., 508  
 Ellison, K. A., 278  
 Elsas, L. J., II, 998  
 Emanuel, B. S., 924  
 Engel, E., 15  
 Erbe, R. W., 1077  
 Erickson, R. P., 879 (BR)  
 Erlich, H. A., 371  
 Erren, M., 1115  
 Escallon, C. S., 700  
 Espinosa, R., III, 1243  
 Estivill, X., 404, 1140 (L)  
 Evans, G. A., 1203  
 Evett, I. W., 869 (L)  
 Faà, 422  
 Fahn, S., 619  
 Fajans, S. S., 607  
 Falls, K. F., 607  
 Farrall, M., 270  
 Farrar, G. J., 590, 634  
 Farrer, L. A., 528, 536, 646 (L), 653 (L), 902  
 Faustinella, F., 1275  
 Feigenbaum, A., 852  
 Feingold, J., 870 (L), 1308  
 Fellous, M., 1008  
 Ferguson-Smith, M. A., 317  
 Ferrell, R. E., 145, 236 (L)  
 Fex, J., 902  
 Fill, C. P., 278  
 Fioretti, G., 781  
 Fletcher, J., 1018  
 Flynn, G. A., 520  
 Foy, C., 902  
 Francis, M. J., 520  
 Francke, U., 725, 988, 1218  
 Fraser, F. C., 874 (L)  
 Frasier, F., 1203  
 Friedman, J. M., 1129  
 Friedman, L., 1235  
 Friedman, T. B., 902  
 Friend, K., 448 (L)  
 Frith, C. D., 536  
 Fujimori, M., 399  
 Fujita, R., 559  
 Funke, H., 1115  
 Gal, A., 876 (L)  
 Gaona, A., 1140 (L)

- Gardner, R. J. M., 1137 (L)  
 Gaskell, C. P., 1211  
 Gasparini, P., 1140 (L)  
 Gatti, R. A., 1343 (L)  
 Gawinowicz, M., 1027  
 Gazit, E., 222  
 Gedeon, A., 968  
 Geller, L. N., 1343 (L)  
 Gerdes, A.-M., 1012  
 Giacalone, J. P., 725  
 Gibson, D. E., 1340 (L)  
 Giebel, L. B., 261  
 Gilchrist, J. H., 896, 1211  
 Giles, A. R., 199  
 Gilgenkrantz, S., 981  
 Gill, T. J., III, 1 (E)  
 Giménez, F. J., 404  
 Ginzinger, D. G., 826  
 Giraldez, R. A., 700  
 Girbau, E., 404  
 Glaser, T., 56  
 Glover, T. W., 1354 (BR)  
 Golbus, M. S., 42  
 Goldgar, D. E., 598  
 Goldstein, S., 451 (BR)  
 Golla, A., 1151  
 Goodfellow, P. J., 1129  
 Goodman, B. K., 1281  
 Goodman, D. S., 1027  
 Goodship, J., 1340 (L)  
 Gorski, S. M., 1129  
 Graeber, M. B., 808  
 Graham, C., 1185  
 Granell, R., 1022  
 Gray, M. R., 331  
 Green, P., 440 (L)  
 Greenberg, D. A., 1053  
 Greenberg, F., 15, 544  
 Greenberg, J., 902  
 Greenhaw, G. A., 677  
 Greenwood, B. M., 411  
 Griffin, L. D., 998  
 Grimm, T., 164, 651 (L), 1341 (L)  
 Grody, W. W., 1281  
 Grompe, M., 483  
 Groth, D. M., 576  
 Growden, J. H., 646 (L)  
 Grundfast, K. M., 902  
 Guarino, E., 781  
 Guenther, C., 1235  
 Gusella, J. F., 551, 619, 896  
 Gustashaw, K. M., 914  
 Haan, E. A., 448 (L), 576  
 Hafez, M., 544  
 Haines, J. L., 551, 896  
 Hall, J. M., 1235  
 Halvorson, S., 1333 (L)  
 Hamsten, A., 208  
 Hanauer, A., 981  
 Harding, A. E., 629  
 Harding, R. M., 411  
 Hardy, J., 648 (L)  
 Harley, H. G., 651 (L), 1341 (L)  
 Harper, P. S., 460 (Op), 651 (L), 1341 (L)  
 Hart, P. S., 126  
 Hauck, W. W., 6  
 Hayasaka, K., 655 (L)  
 Hayden, M. R., 382  
 Hayflick, S., 834  
 Haynes, C. S., 1211  
 Hayward, C., 607, 749  
 Hebert, A., 677  
 Hecht, F., 242 (BR)  
 Hecht, J. T., 677  
 Hecker, K., 758  
 Hedrick, A., 382  
 Helliwell, T. R., 508  
 Helms, C., 1267  
 Hennies, H.-C., 1031  
 Hertz, J. M., 15, 1291  
 Hill, A., 1185  
 Hill, A. V. S., 411  
 Hillen, D., 968  
 Hirata, Y., 1018  
 Ho, M. F., 317  
 Hoar, D. I., 544  
 Hodge, S. E., 1053  
 Holder, S., 270  
 Holloway, S., 749  
 Holme, E., 360  
 Holmes, S. A., 261  
 Holt, I. J., 629  
 Holtzman, N. A., 457 (E)  
 Honeyman, J., 1340 (L)  
 Hopwood, J. J., 787  
 Horn, M., 876 (L)  
 Horn, N., 1012  
 Horton, W. A., 677  
 Hoth, C., 902  
 Howell, N., 443 (L), 447 (L), 1333 (L)  
 Howells, R., 1342 (L)  
 Huggins, R. M., 1067  
 Humphries, M. M., 634  
 Humphries, P., 590, 634  
 Humphries, S. E., 208  
 Hunt, P. A., 1162  
 Hymes, J., 126  
 Ii, S., 29  
 Iles, D., 1151  
 Inglehearn, C. F., 590  
 Ishimura-Oka, K., 1275  
 Jackson, C. E., 795, 1350 (L)  
 Jacobs, K. H., 1018  
 Jacobson, D. R., 195  
 James, K., 1104  
 Janes, S. R., 1077  
 Jaspers, N. G. J., 1343 (L)  
 Jaubert, F., 1008  
 Jay, M., 590  
 Jeanpierre, M., 960  
 Jenkins, D., 1018  
 Jenkins, T., 107, 1301  
 Jensen, F. S., 1086  
 Jin, W.-D., 795  
 Johns, D. R., 872 (L)  
 Johnson, A., 15  
 Johnson, J. P., 882 (BR)  
 Johnson, K. J., 1151  
 Johnsson, V., 816  
 Johnston, H., 1348 (L)  
 Jones, C., 56  
 Jordan, S. A., 634  
 Kaback, M., 438 (L)  
 Kakulas, B. A., 576  
 Kalitsis, P., 706, 717  
 Kamboh, M. I., 145, 236 (L)  
 Kan, Y. W., 422  
 Kang, S.-S., 1281  
 Kaplan, J., 981  
 Kapsa, R. M. I., 1337 (L)  
 Kaspar, F., 1318  
 Kawaguchi, H., 766  
 Kay, A. C., 394  
 Kazantsev, A., 1211  
 Kazazian, H. H., Jr., 15  
 Kearns, W. G., 700

- Keats, B., 559  
 Keen, T. J., 590  
 Kendler, K. S., 1259  
 Kenna, P., 634  
 Kerem, B.-s., 222  
 Kerem, E., 222  
 Kern, R. M., 1281  
 Kessling, A., 92  
 Kiely, D. K., 528  
 Kihara, S., 1275  
 Kim, H.-S., 842  
 King, M.-C., 234 (Obit), 515, 1231, 1235  
 Klein, C. J., 950  
 Klein, D., 1281  
 Klein, J., 766  
 Klitz, W., 371  
 Klocker, H., 1318  
 Kneebone, C. S., 576  
 Koch, M. C., 651 (L), 1341 (L)  
 Koenig, M., 559  
 Kohn, M. A., 476  
 Komatsu, K., 655 (L)  
 Koprivnikar, K., 303  
 Korenberg, J. R., 294  
 Kostyu, D. D., 6  
 Kowbel, D., 382  
 Kramer, P. L., 619  
 Krawczak, M., 652 (L)  
 Kremer, E., 968  
 Kristidis, P., 1178  
 Kruse, T. A., 492, 1012  
 Kubacka, I., 1333 (L)  
 Kuhl, W., 394  
 Kumar-Singh, R., 634  
 Kunkel, L. M., 576  
 Kupke, K. G., 808  
 Kuwano, A., 182  
 Kwiatkowski, D., 411  
 Kwiatkowski, D. J., 619  
 Kwitrovich, P. O., Jr., 1038  
  
 Laca, Z., 15  
 Lacerra, G., 781  
 La Du, B. N., 1086, 1104  
 Laing, N. G., 576  
 Laird, C. D., 1171  
 Lam-Po-Tang, P. R. L., 1348 (L)  
 Landsberger, D., 427  
 Lange, E., 1343 (L)  
 Lange, K., 859, 1343 (L)  
  
 Langley, S. D., 998  
 Larsson, N.-G., 360  
 Lastra, A. A., 1211  
 Laxer, G., 208  
 Layton, M. G., 576  
 Lázaro, C., 404  
 LeBeau, M. M., 1243  
 Lebo, R. V., 42  
 Ledbetter, D. H., 182, 690  
 Ledbetter, S. A., 182  
 Ledley, F. D., 1353 (BR)  
 Lee, M. K., 1235  
 Lehmann-Horn, F., 1151  
 Lehrach, H., 317  
 Leigh, D., 1348 (L)  
 Leitersdorf, E., 427  
 LeMaire, R., 1162  
 Lenoir, G., 364  
 Leonard, J. V., 1281  
 Lerer, I., 1185  
 Lertrit, P., 1337 (L)  
 Lester, D. H., 590  
 Levy, Y., 427  
 Lewis, R. A., 690  
 Lidral, A. C., 567  
 Lillicrap, D., 199  
 Lin, K.-H., 237 (L)  
 Lin, K.-S., 237 (L)  
 Lin, L.-I., 237 (L)  
 Lindgren, V., 988  
 Lindsey, J., 590  
 Liston, W. A., 749  
 Livingstone, J., 749  
 Lockridge, O., 1086  
 Loesch, D. Z., 968, 1067  
 Longo, N., 998  
 Lorenz, B., 876 (L)  
 Lubrano, T., 1086  
 Ludwig, M., 164  
 Lukka, M., 816  
 Lupski, J. R., 690  
 Lussier-Cacan, S., 92  
 Lynch, E. D., 42  
 Lynch, M., 968  
  
 McClatchey, A. I., 896  
 McClure, G., 371  
 McCourt, P. A. G., 787  
 McCullough, D. A., 443 (L), 1333 (L)  
 McDonald, B., 1348 (L)  
  
 MacDougall, M., 190  
 McElreavey, K., 1008  
 McGinniss, M. J., 15  
 McInnis, M. G., 544  
 McKeithan, T. W., 1243  
 McKenna-Yasek, D., 896  
 Mackey, D., 1333 (L)  
 Mackie, A., 607  
 McKusick, V. A., 663 (CHG-PA)  
 MacLean, C. J., 1259  
 MacLennan, D. H., 1151  
 McMichael, A. J., 411  
 McQuillan, C., 717  
 Maestrini, E., 156  
 Magallón, M., 434  
 Mahuran, D. J., 1046  
 Maiste, P. J., 1139 (L)  
 Majda, B. T., 576  
 Majumder, P. P., 1328  
 Manass, S., 1348 (L)  
 Mandel, J. L., 559  
 Marazita, M., 902  
 Marescau, B., 1281  
 Margaritte, P., 1231  
 Markiewicz, D., 1178  
 Martin-Gallardo, A., 278  
 Martin-Villar, J., 434  
 Marzuki, S., 1337 (L)  
 Masciangelo, F., 781  
 Mathieu, M., 981  
 May, E., 870 (L)  
 Meindl, A., 1151  
 Meiner, V., 427  
 Meitinger, T., 1151  
 Melki, J., 892  
 Meloni, A., 422  
 Mendell, J. R., 950  
 Menninger, J., 71  
 Mérette, C., 515  
 Metaxotou, C., 544  
 Metzger, A. K., 584  
 Mijovic, C. H., 1018  
 Mikkelsen, M., 15, 544  
 Milatovich, A., 1218  
 Miller, C. S., 834  
 Miller, O. J., 826  
 Milunsky, A., 643 (L), 902  
 Mishra, S. K., 1267  
 Mitchell, D. A., 137  
 Miyake, Y., 498  
 Mokini, V., 875 (L)



- Molano, J., 1022  
 Molloy, C. M., 1195  
 Monaco, A. P., 317  
 Montag, E., 498  
 Moraes, C. T., 934  
 Morell, R., 902  
 Mori, M., 56  
 Morris, G. E., 508  
 Morrison, K. E., 520  
 Motulsky, A. G., 881 (BR)  
 Mountford, R. C., 508  
 Mulder, L., 1308  
 Mules, E. H., 834  
 Mullan, M., 648 (L)  
 Müller, B., 892  
 Müller, U., 808  
 Muller, V. J., 787  
 Mulley, J. C., 576, 968  
 Munnich, A., 364  
 Murray, E. W., 199  
 Murray, J., 190  
 Myers, R. H., 528, 646 (L),  
 653 (L)  
 Myers, R. M., 1218  
  
 Nagel, R. L., 71  
 Najfeld, V., 71  
 Nakamura, Y., 56, 65, 399  
 Nance, W. E., 76, 902  
 Narcy, P., 364  
 Nash, E., 1185  
 Nath, S. K., 1328  
 Nathans, J., 498  
 Natowicz, M. R., 465, 476  
 Navidi, W., 347  
 Neale, M. C., 76  
 Neilly, M. E., 1243  
 Nelen, M., 801  
 Newton, V., 902  
 Nichols, B., 567  
 Noer, A. S., 1337 (L)  
 Notarangelo, L. D., 156  
 Novelli, G., 875 (L), 1140 (L)  
 Nunes, V., 1140 (L)  
 Nussbaum, R. L., 742  
 Nwankwo, M., 145  
  
 Ober, C., 6  
 O'Brien, W. E., 439 (L)  
 O'Connell, P., 42, 842  
 O'hUigin, C., 766  
  
 Oka, K., 1275  
 Okui, K., 65  
 Oldfors, A., 360  
 Olek, K., 164  
 Oniki, R. S., 598  
 Oosterwijk, J. C., 801  
 Oranje, A. P., 801  
 Oshimura, M., 65  
 Otani, T., 1018  
 Ott, J., 278, 515  
 Oudet, C., 981  
 Ouellette, S., 92  
 Ozcelik, T., 988  
 Ozelius, L. J., 619  
  
 Pagano, L., 781  
 Pangalos, C., 544  
 Parenti, G., 483  
 Park, J. W., 56  
 Park, V. M., 914  
 Parolini, O., 156  
 Patel, P. I., 690  
 Patil, S. R., 15  
 Pawlowitzki, I. H., 1195  
 Peacock, R., 208  
 Pearson, P. L., 700  
 Peltonen, L., 816  
 Penny, M. A., 1018  
 Pentao, L., 690  
 Percy, A. K., 278  
 Pericak-Vance, M. A., 896,  
 1211, 1218  
 Petersen, M. B., 15, 544  
 Petrova-Benedict, R., 852  
 Petterson, A., 1012  
 Phillips, H., 576  
 Phillips, J. A., III, 871 (L)  
 Piazza, A., 875 (L)  
 Pignatti, P. F., 1140 (L)  
 Pinckers, A. J. L. G., 1195  
 Piussan, C., 981  
 Ploughman, L. M., 1259  
 Poncin, J., 981  
 Pongratz, D., 1151  
 Potter, H., 1343 (L)  
 Poulton, J., 651 (L)  
 Prchal, J., 394  
 Pritchard, C., 1218  
 Pritchard, M., 968  
 Puck, J. M., 742  
 Purohit, K., 559  
  
 Purvis-Smith, S. G., 1348 (L)  
 Pyeritz, R. E., 84  
  
 Qian, J. F., 870 (L)  
  
 Radmayr, C., 1318  
 Radosavljevic, D., 896  
 Rafi, M. A., 1252  
 Ragno, P., 492  
 Ramesar, R., 902  
 Ramsay, M., 1301  
 Raoul, O., 544  
 Raskind, W., 896  
 Rassool, F. V., 1243  
 Ray, P. N., 950  
 Raymond, J.-P., 1008  
 Read, A. P., 902  
 Reed, J. K., 758  
 Reilly, P., 640 (ASHG)  
 Reis, A., 1031  
 Renges, H.-H., 208  
 Reshef, A., 427  
 Reynolds, L. W., 834  
 Riccardi, V. M., 1144 (BR)  
 Ricci, E., 934  
 Richards, R. I., 448 (L), 968  
 Richaud, F., 1008  
 Ridley, R. M., 536  
 Riess, O., 382  
 Risch, N., 619, 653 (L)  
 Rivella, S., 156  
 Rivlin, Y., 222  
 Robinson, B. H., 852  
 Rocchi, M., 156  
 Roe, C. R., 229  
 Roldan, A. L., 492  
 Romero, N., 364  
 Rommens, J., 1178  
 Ropers, H.-H., 1195  
 Rosatelli, M. C., 422  
 Rosenberg, C., 700  
 Rosenfield, J. M., 1077  
 Rosenstein, B. J., 1185  
 Roses, A. D., 1211, 1218  
 Rosseneu, M., 208  
 Rothschild, C. B., 607  
 Rothstein, M. A., 457 (E)  
 Rötig, A., 364  
 Roulson, D., 544  
 Rubinstein, H. M., 1086  
 Rudd, N., 852

- Rudd, N. L., 544  
Rustin, P., 364  
Rüther, K., 1195  
Rybicki, A. C., 71
- Saba, L., 422  
Sabl, J. F., 1171  
Saha, N., 208  
Saito, S., 65  
Sajantila, A., 816  
Sakemi, K., 655 (L)  
Salido, E. C., 303  
Salvati, F., 781  
San Agustin, T. B., 902  
Sanal, O., 1343 (L)  
Sangiulio, F., 875 (L)  
Santachiara-Benerecetti, S., 156  
Saraiva, M. J. M., 1027  
Sardu, R., 422  
Sarkar, S., 1328  
Sato, W., 655 (L)  
Saudubray, J.-M., 364  
Sauvage, P., 870 (L)  
Saville, T., 1348 (L)  
Sawaishi, Y., 655 (L)  
Scarallo, A., 781  
Scharf, S. J., 371  
Schieken, R. M., 76  
Schimke, R. N., 450 (BR)  
Schindelbauer, D., 1151  
Schinzel, A. A., 15, 288, 544, 981  
Schofield, I., 1340 (L)  
Schon, E. A., 934  
Schuback, D. E., 619  
Schuchman, E. H., 795  
Schulte, H., 1115  
Schulte, M., 1115  
Schwartz, R. S., 71  
Schwartz, S., 174, 544  
Selinger, E., 92  
Seret, H., 222  
Sertedaki, A., 590  
Shaffer, L. G., 717  
Shapiro, L. J., 303  
Sharp, E. M., 634  
Sheehy, M. J., 607  
Sheffield, V. C., 567, 584  
Sherman, S., 1137 (L)  
Sherman, W., 1027  
Shields, G. F., 758
- Shih, L. Y., 137  
Shiloh, Y., 1343 (L)  
Shinzato, K., 498  
Shoji, Y., 655 (L)  
Shoshani, T., 222  
Shridhar, V., 826  
Sirugo, G., 559  
Skare, J., 902  
Slaugenhaupt, S. A., 544  
Slavkin, H., 190  
Smith, C., 852  
Smith, D. I., 826  
Smith, L. C., 1275  
Sobel, E., 1343 (L)  
Sobell, J. L., 29  
Solera, J., 434, 1022  
Sommer, S. S., 29  
Sparkes, R. S., 1353 (BR)  
Speer, M. C., 576, 1211  
Spielvogel, H., 1031  
Spritz, R. A., 261  
Spurdle, A., 107, 1301  
Stajich, J. M., 1211  
Steele, M. W., 1351 (L)  
Stetten, G., 15, 700  
Stevens, C. A., 902  
Stewart, C. C., 742  
Stewart, G. D., 551  
Stoll, C., 870 (L)  
Stone, E. M., 567  
Ström, M., 816  
Sudoyo, H., 1337 (L)  
Sutherland, G. R., 968  
Suthers, G. K., 520, 887 (E)  
Sutton, H. E., 658 (BR)  
Sweeney, M. G., 629
- Taggart, R. T., 826  
Takada, G., 655 (L)  
Takiguchi, S., 56  
Tal, A., 222  
Talmud, P., 208  
Tanigami, A., 56  
Tanzi, R. E., 551  
Tatuch, Y., 852  
Taylor, A. M. R., 1343 (L)  
Taylor, C., 411  
Terwilliger, J., 278  
Thomas, G. H., 677, 834  
Thyagarajan, D., 1337 (L)  
Tikkanen, M. J., 208
- Tokino, T., 56, 65  
Toniolo, D., 156  
Tønnesen, T., 1012  
Tranebjaerg, L., 15, 544  
Tribioli, C., 156  
Tripputi, P., 492  
Trofatter, J., 896  
Tryggvason, K., 1291  
Tsipouras, P., 451 (BR)  
Tsui, L.-C., 1178  
Tulinus, M. H., 360  
Tumpson, J. E., 84  
Turner, G., 968  
Tzourio, C., 645 (L)
- Überreiter, S., 1318  
Uchigata, Y., 1018
- Vance, J. M., 1211, 1218  
Van den Bergh, P., 896  
van den Hurk, J. A. J. M., 1195  
van de Pol, T. J. R., 1195  
van der Spek, A. F. L., 1086  
van der Westhuyzen, D. R., 427  
van Melle, E., 1243  
van Ommen, G. J. B., 317  
van Oost, B. A., 801  
van Osch, L. D. M., 801  
van Zandvoort, P. M., 801  
Vázquez, C., 404  
Vilain, E., 1008  
Vissel, B., 706  
Vnencak-Jones, C. L., 871 (L)  
Voevoda, M. I., 758  
von Eckardstein, A., 1115
- Wagner, R. G., Jr., 902  
Wang, W., 1046  
Ward, D. C., 71  
Warren, A. C., 544  
Washington, S. S., 1031  
Waterman, M. S., 347  
Wathen, T. M., 914  
Watkins, P. C., 551  
Weaver, R., 1267  
Weber, B., 382  
Weber, J. L., 1211, 1235  
Weeks, D. E., 859  
Weemaes, C. M. A., 1343 (L)  
Wei, J. T., 137  
Weir, B. S., 869 (L), 1139 (L)

Weitz, C. J., 498  
Wells, S. A., Jr., 399  
Wenger, D. A., 1252  
Went, L. N., 498  
Wertz, D. C., 1077  
Wexler, N. S., 551  
Wherret, J., 852  
White, S. J., 576  
Wieringa, B., 1195  
Wilcox, E. R., 902  
Wilfond, B., 438 (L)  
Wilton, S. D., 576  
Winship, I., 902  
Wissman, P. B., 1281  
Wittebol-Post, D., 801

Wolf, B., 126  
Wolff, D. J., 174  
Wolman, S. R., 800 (BR)  
Worthen, H. G., 896

Xhignesse, M., 92  
Xie, B., 1046  
Xu, C.-F., 208

Yaar, L., 222  
Yager, R. M., 137  
Yahav, Y., 222  
Yamaoka, L. H., 1211  
Yandell, D. W., 371  
Yang, B.-Z., 229

Yang-Feng, T. L., 988  
Yates, J., 483  
Yen, P. H., 303  
Yip, M.-Y., 1348 (L)  
Yu, L.-C., 303  
Yu, S., 968

Zeichner-David, M., 190  
Zhang, X.-L., 1252  
Zhou, J., 1291  
Zhu, N., 1218  
Zoghbi, H., 278  
Zrenner, E., 498, 1195  
Zuo, J., 1218



## Subject Index to Volume 50

(ASHG) = American Society of Human Genetics report; (BR) = book review; (CHG-PA) = Congress of Human Genetics presidential address; (CHG-PL) = Congress of Human Genetics plenary lecture; (E) = editorial; (L) = letter to the editor; (Obit) = obituary; (Op) = opinion

- Acyl-CoA dehydrogenase deficiency, medium chain, 229
- Adenomatous polyposis, familial, chromosome 5, deletions, 988
- Affected-pedigree-member method, multilocus extension, 859
- Age at onset
  - Huntington disease, 528
  - X-linked recessive modifier, 536
- Age-at-onset covariate, breast cancer heterogeneity analysis, 515
- Age-at-onset distributions, 652 (L), 653 (L)
- $\alpha$ -5(IV)Collagen chain, juvenile-onset Alport syndrome, 1291
- $\alpha$ -L-iduronidase, immunoquantification and enzyme kinetics, mucopolysaccharidosis type I, 787
- $\alpha$ -Methylmannoside, N-acetylglucosamine 1-phosphotransferase activity, 137
- $\alpha$ -Subunit, hexosaminidase A, mutations, 834
- Alport syndrome, juvenile onset,  $\alpha$ -5(IV)collagen chain, 1291
- Alzheimer disease, 648 (L)
  - Maternal age and somatic mosaicism, 1342 (L), 1343 (L)
  - Segregation analysis, 645 (L), 646 (L)
- Amelogenesis, enamel protein gene, 303
- Amphidicolin-inducible fragile-site expression, 76
- Amyloid cardiomyopathy, transthyretin mutation, 1027
- Androgen receptor, point mutation, Reifstein syndrome, 1318
- Antigen group (x/y) polymorphism, apo B, 208
- Antigens, HLA class I, interethnic genetic differentiation, 411
- Apolipoprotein A-I-containing lipoproteins, apo A-IV gene nonsynonymous polymorphic sites, 1115
- Apolipoprotein A-IV, nonsynonymous polymorphic sites, 1115
- Apolipoprotein AI-CIII-AIV, and lipid and lipoprotein traits, 92
- Apolipoprotein B
  - Antigen group (x/y) polymorphism, 208
  - Lipid and lipoprotein traits, 92
  - Lipoproteins containing, apo A-IV gene nonsynonymous polymorphic sites, 1115
  - Pedigree and sib-pair linkage analysis, 1038
- Apolipoprotein E genotype, dietary and plasma cholesterol, 236 (L)
- Arginase deficiency, 1281
- Arylsulfatase B gene mutations, mucopolysaccharidosis type VI, 795
- American Society of Human Genetics, genetics and privacy, 640 (ASHG)
- Ataxia
  - Cerebellar, mitochondrial genome duplication, 364
  - Friedreich, founder effect, 559
- Ataxia-telangiectasia, genetic heterogeneity, 1343 (L)
- $\beta$ -Thalassemia
  - Molecular characterization, 422
  - 32(C-A) mutant, 237 (L)
- Biopsy, skin, 875 (L)
- Biotinidase, serum, deficiency, 126
- Blood groups, discovery, 671 (CHG-PL)
- Blood infection, malaria, segregation analysis, 1308
- Breakpoint, t(14q21q) Robertsonian translocations, 717
- Breast cancer
  - Familial, 17q21, 1231
  - Gene, 17q, 1235
  - Heterogeneity analysis, age at onset, 515
- Butyrylcholinesterase
  - J-variant, DNA mutations, 1104
  - K-variant, DNA mutation, 1086
- c-kit proto-oncogene, piebaldism, 261
- Cancer, breast
  - Familial, 17q21, 1231
  - Gene, 17q, 1235
  - Heterogeneity analysis, age at onset, 515
- Cardiomyopathy, amyloid, transthyretin mutation, 1027
- Carrier detection/screening
  - Cystic fibrosis, 439 (L)
  - Duchenne muscular dystrophy, 448 (L)
- Caucasian genes, American blacks, 145
- Cerebellar ataxia, mitochondrial genome duplication, 364
- Charcot-Marie-Tooth type 1A, markers, 42

- Cholesterol, dietary and plasma, apo E genotype, 236 (L)
- Cholesterol ester transfer protein gene, and lipid and lipoprotein traits, 92
- Choroideremia candidate gene, point mutations, 1195
- Chromosome 1, autosomal dominant nemaline myopathy, 576
- Chromosome 2, Waardenburg syndrome type I, 902
- Chromosome 3p14, fragile site, 1243
- Chromosome 3p21.3, DNF15S2 and D3S94, 826
- Chromosome 4p16, DNA markers, recombination, Huntington disease, 1218
- Chromosome 5
- Deletions, familial adenomatous polyposis, 988
  - Spinal muscular atrophy, 520
- Chromosome 5q
- Autosomal dominant limb-girdle muscular dystrophy, 1211
  - Proximal spinal muscular atrophy types II and III, 892
- Chromosome 6, mapping markers, 65
- Chromosome 9q34, torsion dystonia, 619
- Chromosome 11, mapping markers, 56
- Chromosome 14
- Maternal uniparental isodisomy, 690
  - Satellite III DNA subfamily, 706
- Chromosome 15q15, erythrocyte protein 4.2, 71
- Chromosome 17p13, lissencephaly, 182
- Chromosome 17q21, familial breast cancer, 1231
- Chromosome 17p, medulloblastoma tumorigenesis, 584
- Chromosome 17q, breast cancer gene, 1235
- Chromosome 19q13.1-q13.2, urokinase-type plasminogen activator receptor gene, 492
- Chromosome 21
- Map, 551
  - Ring formation, 15
- Chromosome 22q11, deletions, DiGeorge syndrome, 924
- Chromosome X
- Enamel protein gene, amelogenin, 303
  - Inactivation, RFLPs, 156
  - Markers, Rett syndrome, 278
  - T-cell, inactivation, 742
- Chromosome Xp21, McLeod locus, 317
- Chromosome Xp21.2-p22.2, keratosis follicularis spinulosa decalvans, 801
- Chromosome Xp22.1-p22.2, Coffin-Lowry syndrome, 981
- Chromosome Xq12-q21.1, dystonia-Parkinsonism syndrome locus, 808
- Chromosome Xq28 CpG islands, RFLPs, 156
- Chromosome Y
- Enamel protein gene, amelogenin, 303
  - Probe p49a, *PvuII* and *TaqI* haplotypes, 107
- Chromosome painting, reciprocal translocations, 700
- Chylomicronemia, familial, lipoprotein lipase gene missense mutation, 1275
- Cleft lip/palate
- Bilateral sporadic, 870 (L)
  - Familial recurrence pattern, 270
- Cleft palate, X linked, *PGK1* and *DXYS1*, 1129
- Clinical genetic services, economics, 84, 1351 (L)
- Cockayne syndrome, phenotype, 677
- Coffin-Lowry syndrome, chromosome Xp22.1-p22.2, 981
- Collagen chain,  $\alpha$ -5(IV), juvenile-onset Alport syndrome, 1291
- CpG islands, Xq28, 156
- Crossing-over, proline-rich protein genes *PRB1* and *PRB2*, 842
- Cys<sub>138</sub>-to-Arg substitution, *G<sub>M2</sub>* activator protein, *G<sub>M2</sub>* gangliosidosis, 1046
- Cystic fibrosis
- Carrier screening, 439 (L)
  - Caucasians, 1185
  - $\Delta F508$  mutation, 404, 875 (L)
  - Exocrine pancreatic function, 1178
  - Gene, nonframeshift deletion, 1022
  - Nonsense mutation, 222
  - Population screening, 438 (L)
  - Prenatal diagnosis, family decision making, 1077
  - Transmembrane regulator gene, frameshift mutation, 1140 (L)
- Cytochrome P450c21 gene, evolutionary origin, 766
- D1S80 locus, forensic casework, 816
- D3S94, chromosome 3p21.3, 826
- D13S71, microsatellite, 1031
- Data base, mtDNA, 1333 (L)
- Deletion(s)
- Chromosome 5, familial adenomatous polyposis, 988
  - Chromosome 22q11, DiGeorge syndrome, 924
  - Codon 255/256, rhodopsin gene, autosomal dominant retinitis pigmentosa, 876 (L)
  - Nonframeshift, cystic fibrosis gene, 1022
  - Prion protein gene, heteroduplexes, 871 (L)
  - X/autosome translocation, 725

- Deletion/insertion, factor IX<sub>Madrid 2</sub>, 434  
 Deletion pedigree, Duchenne muscular dystrophy, 448 (L)  
 $\Delta F508$  mutation, cystic fibrosis, 404, 875 (L)  
 Dentin phosphoprotein gene, dentinogenesis types II and III, 190  
 Dentinogenesis types II and III, dentin phosphoprotein gene, 190  
 Diabetes, maturity-onset, linkage analysis, 607  
 Diabetes mellitus  
   Insulin dependent, inheritance, 1018  
   Mitochondrial genome duplication, 364  
 Diagnosis, prenatal  
   Cystic fibrosis, family decision making, 1077  
   mtDNA<sup>8993 T-G</sup> disease, 629  
   Diagnostic status, lod-score sensitivity, 1053  
   DiGeorge syndrome, etiology, 924  
   Disequilibrium, linkage, estimation, 1139 (L)  
   Disomy 15, uniparental, trisomy 15, 1348 (L)  
   DNA fingerprinting, population genetics, 440 (L), 441 (L)  
   DNA property rights, 869 (L)  
   DNF15S2, chromosome 3p21.3, 826  
 Down syndrome  
   Heart disease and duodenal stenosis, 294  
   Nondisjunction, 288  
 Duchenne muscular dystrophy  
   Carrier detection, 448 (L)  
   Somatic reversion/suppression, 950  
 Duodenal stenosis, Down syndrome, 294  
 DXYS1, X-linked cleft palate, 1129  
 Dystonia-Parkinsonism syndrome locus, flanking markers, 808  
 Dystrophin, C-terminal domains, muscle membrane, 508  
 DYT1 gene, chromosome 9q34, 619  
  
 Economics, clinical genetics services, 84, 1351 (L)  
 Eighth International Congress of Human Genetics, presidential address, 663 (CHG-PA)  
 Enamel protein gene, amelogenin, 303  
 Encephalopathy, mitochondrial myopathy, lactic acidosis, strokelike episodes, 655 (L)  
 Enzyme kinetics,  $\alpha$ -L-iduronidase, mucopolysaccharidosis type I, 787  
 Epigene conversion, gene mapping, 1171  
 Erythrocyte protein 4.2 gene, mapping, 71  
 Eugenics, 457 (E)  
 Exclusion map, pre-eclampsia, 749  
 Exocrine pancreatic function, cystic fibrosis, 1178  
  
 Factor IX gene  
   Deletion/insertion, 434  
   Mutations, parental origin, 164  
 Factor IX<sub>Madrid 2</sub>, deletion/insertion, 434  
 Fecundability, and HLA-DR, 6  
 Finger ridge count, fragile-X families, 1067  
 Fisher, R. A., 671 (CHG-PL)  
 Forensic casework, D1S80 locus, 816  
 Founder effect, Friedreich ataxia, 559  
 Fragile site, 3p14, 1243  
 Fragile-site expression, amphidicilin inducible, 76  
 Fragile X, finger ridge count, 1067  
 Fragile-X syndrome, heritable unstable element, 968  
 Frameshift mutation, cystic fibrosis transmembrane regulator gene, 1140 (L)  
 Frequency calculation, accuracy, 874 (L)  
 Friedreich ataxia, founder effect, 559  
  
 Gangliosidosis, G<sub>M2</sub>, AB variant, G<sub>M2</sub> activator protein, Cys<sub>138</sub>-to-Arg substitution, 1046  
 Gene transfer, Recombinant DNA Advisory Committee, 245 (Op)  
 Genetic discrimination, 457 (E), 476  
   Law, 465 (R)  
 Genetic laboratory services, 643 (L)  
 Genotyping PCR, multiple-tubes approach, 347  
 Germ-line mosaicism, valine-to-methionine substitution, type IIB von Willebrand disease, 199  
 Glucose-6-phosphate dehydrogenase, polymorphisms, 394  
 Glycoprotein IIb-binding domain, von Willebrand factor, germ-line mosaicism, 199  
 G<sub>M2</sub> activator protein, Cys<sub>138</sub>-to-Arg substitution, G<sub>M2</sub> gangliosidosis AB variant, 1046  
 G<sub>M2</sub> gangliosidosis AB variant, G<sub>M2</sub> activator protein, Cys<sub>138</sub>-to-Arg substitution, 1046  
 Growth factor alpha, transforming, genetic variation, 870 (L)  
  
 Haldane, J. B. S., 671 (CHG-PL)  
 Haplotype(s)  
   Friedreich ataxia, founder effect, 559  
   PvuII and TaqI, Y chromosome probe p49a, 107  
 Hb Lepore-Boston gene, origin heterogeneity, 781  
 Heart disease, congenital, Down syndrome, 294  
 Heteroduplexes, prion protein gene deletions, 817 (L)  
 Heterogeneity  
   Allelic, hyperkalemic periodic paralysis and paramyotonia congenita, 896  
   Genetic  
     Ataxia-telangiectasia, 1343 (L)

- Limb-girdle muscular dystrophy, 1211
- Malignant hyperthermia susceptibility, 1151
- Locus, 1259
- Maturity-onset diabetes, 607
- Mucopolysaccharidosis type VI, 795
- Origin, Hb Lepore-Boston gene, 781
- Phenotypic, 887 (E)
- Heterogeneity analysis, breast cancer, 515
- Hexosaminidase A,  $\alpha$ -subunit, mutations, 834
- HLA class I antigens, interethnic genetic differentiation, 411
- HLA-DR, and fecundability, 6
- Huntington disease
  - Age at onset
    - Paternal age, 528
    - X-linked recessive modifier, 536
  - 4p16 DNA markers, recombination, 1218
  - Genetics abuse, 460 (Op)
  - Polymorphic DNA markers, 382
- Hybridization
  - Fluorescence in situ, Robertsonian translocations, 174
  - Multicolor in situ, CMT1A, markers, 42
- Hypercholesterolemia, familial, LDL receptor gene nonsense mutation, 427
- Hyperkalemic periodic paralysis, allelic heterogeneity, 896
- Hyperthermia susceptibility, malignant, genetic heterogeneity, 1151
- Ichthyosis, X-linked, steroid sulfatase gene point mutations, 483
- Identity-by-descent method, mapping quantitative traits, 598
- Immunodeficiency, X-linked combined, carriers, 742
- Immunoquantification,  $\alpha$ -L-iduronidase, mucopolysaccharidosis type I, 787
- Imprinting, genetic, Huntington disease, 528
- Inactivation
  - Chromosome X, RFLPs, 156
  - T-cell X, maximum-likelihood analysis, 742
- Infection, blood, malaria, segregation analysis, 1308
- Insertion/deletion, factor IX<sup>Madrid 2</sup>, 434
- Insulin resistance, insulin-receptor gene nonsense mutation, 998
- Insulin-dependent diabetes mellitus, inheritance, 1018
- Insulin-receptor gene, nonsense mutation, insulin resistance, 998
- Interstitial telomeric sequences, 914
- Isodisomy, maternal uniparental, chromosome 14, 690
- Kearns-Sayre syndrome, deleted mtDNA transmission, 360
- Keratosis follicularis spinulosa decalvans, linkage analysis, 801
- Lactic acidosis, mitochondrial (encephalo)myopathy, strokelike episodes, 655 (L), 934
- Law, genetic discrimination, 465 (R)
- LDL receptor gene, nonsense mutation, familial hypercholesterolemia, 427
- Leber (hereditary) optic neuropathy, 443 (L), 446 (L), 447 (L)
  - ND-1 mutation, 872 (L)
- Leigh disease, T $\rightarrow$ G, 852
- Limb-girdle muscular dystrophy, genetic heterogeneity, 1211
- Linkage analysis
  - Affected-pedigree-member method, 859
  - CMT1A, markers, 42
  - Keratosis follicularis spinulosa decalvans, 801
  - Maturity-onset diabetes, 607
  - Menkes disease, 1012
  - Multilocus, exclusion mapping, Rett syndrome, 278
  - Multipoint, mapping quantitative traits, 598
  - Pedigree and sib-pair, apo B gene, 1038
- Linkage disequilibrium, estimation, 1139 (L)
- Lipid traits, apo B, apo AI-CIII-AIV, cholesterol ester transfer protein gene, 92
- Lipoprotein lipase gene, missense mutation, familial chylomicronemia, 1275
- Lipoproteins, apo B- and apo A-I-containing, apo A-IV gene nonsynonymous polymorphic sites, 1115
- Lipoprotein traits, apo B, apo AI-CIII-AIV, cholesterol ester transfer protein gene, 92
- Lissencephaly, chromosome 17p13, 182
- Lod score sensitivity, diagnostic status, 1053
- Low allele, Y-associated XY275, 1301
- McLeod locus, Xp21, 317
- Major histocompatibility complex, and reproduction, 1 (E)
- Malaria, blood infection, segregation analysis, 1308
- Malignant hyperthermia susceptibility, genetic heterogeneity, 1151
- Map(s)/mapping
  - Chromosome 6, RFLP markers, 65

- Chromosome 11, markers, 56  
 Chromosome 21, 551  
 Erythrocyte protein 4.2 gene, 71  
 Exclusion  
   Multilocus linkage analysis, Rett syndrome, 278  
   Pre-eclampsia, 749  
 Gene, epigene conversion, 1171  
 Heart disease and duodenal stenosis, Down syndrome, 294  
 McLeod locus, Xp21, 317  
 Multiple endocrine neoplasia type 1, 399  
 Quantitative traits, 598  
 Mast/stem cell growth factor receptor proto-oncogene, piebaldism, 261  
 Maternal age, somatic mosaicism and Alzheimer disease, 1342 (L), 1343 (L)  
 Maximum-likelihood analysis, T-cell X inactivation, 742  
 Medium-chain acyl-CoA dehydrogenase deficiency, 229  
 Medulloblastoma tumorigenesis, chromosome 17p, 584  
 Meiosis I and II nondisjunction, recombination, trisomy 21, 1137 (L)  
 Menkes disease, linkage analysis, 1012  
 Metabolism, sulfatide, SAP-1 deficiency, 1252  
 Missense mutation, lipoprotein lipase gene, familial chylomicronemia, 1275  
 Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes, skeletal muscle, 934  
 Mitochondrial marker, Asian-specific region V, 758  
 Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, 655 (L)  
 Mitochondrial tRNA<sup>Leu(UUR)</sup> mutation, MELAS, 934  
 Mosaicism  
   Germ line, valine-to-methionine substitution, type IIb von Willebrand disease, 199  
   Germinal, X-linked diseases, 960  
   Somatic, maternal age and Alzheimer disease, 1342 (L), 1343 (L)  
 mtDNA<sup>8993 T-G</sup> disease, prenatal diagnosis, 629  
 Mucopolysaccharidosis  
   Type I,  $\alpha$ -L-iduronidase, 787  
   Type VI, arylsulfatase B gene mutations, 795  
 Multicolor in situ hybridization, CMT1A, markers, 42  
 Multilocus linkage analysis, exclusion mapping, Rett syndrome, 278  
 Multiple endocrine neoplasia type 1, 399  
 Multiple-tubes approach, genotyping, PCR, 347  
 Muscular atrophy  
   Spinal, locus map, chromosome 5, 520  
   Types II and III, proximal spinal, chromosome 5q, 892  
 Muscular dystrophy  
   Duchenne  
     Carrier detection, 448 (L)  
     Reversion/suppression, 950  
     Limb-girdle, genetic heterogeneity, 1211  
   Myotonic dystrophy  
     Congenital, 651 (L), 651 (L)  
     Genetic risks, 1340 (L), 1341 (L)  
 N-acetylglucosamine 1-phosphotransferase activity, mucopolysaccharidosis II and III, 137  
 ND-1 mutation, Leber hereditary optic neuropathy, 872 (L)  
 Nemaline myopathy, autosomal dominant, chromosome 1, 576  
 Nonallelic mutations, mucopolysaccharidosis II and III, 137  
 Nonconjunction, 1137 (L)  
 Nondisjunction, 1137 (L)  
   Down syndrome, 288  
   Meiosis I and II, recombination, trisomy 21, 1137 (L)  
   Trisomy 21, 544  
 Nonpenetrance, maturity-onset diabetes, 607  
 Nonsense mutation  
   Cystic fibrosis, 222  
   Insulin-receptor gene, insulin resistance, 998  
   LDL receptor gene, familial hypercholesterolemia, 427  
 Nonsynonymous polymorphic sites, apo A-IV gene, 1115  
 Opsin, blue-sensitive, tritanopia, 498  
 Optic neuropathy, Leber (hereditary), 443 (L), 446 (L), 447 (L)  
   ND-1 mutation, 872 (L)  
 p49a, PvuII and TaqI haplotypes, 107  
 Pancreatic function, exocrine, cystic fibrosis, 1178  
 Paramyotonia congenita, allelic heterogeneity, 896  
 Parkinsonism, dystonia, locus, flanking markers, 808  
 Pedigree linkage analysis, apo B gene, 1038  
 Periodic paralysis, hyperkalemic, allelic heterogeneity, 896  
 Peripherin-RDS locus, autosomal dominant retinitis pigmentosa, 634  
 PGK1, X-linked cleft palate, 1129



- Phenotype(s)  
 Mucopolysaccharidosis type VI, 795  
 Sex, testis-determining region, 1008  
 Xeroderma pigmentosum and Cockayne syndrome, 677
- Phenotype heterogeneity, 887 (E)
- Piebaldism, *c-kit* (mast/stem cell growth factor receptor) proto-oncogene, 261
- Plasminogen, urokinase type, activator receptor gene, 492
- Point mutation(s)  
 Androgen receptor, Reifenstein syndrome, 1318  
 Choroideremia candidate gene, 1195  
 Steroid sulfatase gene, X-linked ichthyosis, 483
- Polymorphism(s), 1333 (L)  
 Antigen group (x/y), apo B, 208  
 DNA sequence, 331  
 Glucose-6-phosphate dehydrogenase, 394  
 Multiallele, 567  
 Repeat, *SCN4A* locus, HYPP and PMC, 896  
 Restriction-fragment-length  
 Markers, chromosome 6, 65  
 Xq28 CpG islands, 156
- Population genetics, DNA fingerprinting, 440 (L), 441 (L)
- Population screening, cystic fibrosis, 438 (L)
- PRB2/1* fusion gene, 842
- Pre-eclampsia, exclusion map, 749
- Prenatal diagnosis  
 Cystic fibrosis, family decision making, 1077  
 mtDNA<sup>8993</sup> T-G disease, 629
- Presidential address, Eighth International Congress of Human Genetics, 663 (CHG-PA)
- Prion protein gene deletions, heteroduplexes, 871 (L)
- Privacy, ASHG statement, 640 (ASHG)
- Proline-rich protein genes *PRB1* and *PRB2*, 842
- Proteins, proline rich, *PRB1* and *PRB2*, 842
- Protein 4.2, erythrocyte, mapping, 71
- Proto-oncogene, *c-kit* (mast/stem cell growth factor receptor), piebaldism, 261
- Protoporphyrin, 1203
- Pseudoautosomal region, sex-chromosome pairing, 1162
- Quantitative traits, mapping, 598
- Reciprocal translocations, chromosome painting, 700
- Recombinant DNA Advisory Committee, gene transfer, 245 (Op)
- Recombination  
 4p16 DNA markers, Huntington disease, 1218  
 Meiosis I and II nondisjunction, trisomy 21, 1137 (L)  
 Rhodopsin and D3S47, 590  
 Sex and age, 551
- Recurrence-pattern analysis, familial, cleft lip/palate, 270
- Reifenstein syndrome, androgen receptor point mutation, 1318
- Repeat polymorphisms, *SCN4A* locus, HYPP and PMC, 896
- Reproduction, MHC and MHC-linked genes, 1 (E)
- Restriction-fragment-length polymorphism(s)  
 Markers, chromosome 6, 65  
 Xq28 CpG islands, 156
- Retinitis pigmentosa  
 Autosomal dominant  
 Cosegregation of RP6 and peripherin-RDS locus, 634  
 Rhodopsin gene codon 255/256 deletion, 876 (L)  
 Rhodopsin, recombination, 590
- Retinoblastoma gene VNTR, 371
- Rett syndrome, chromosome X markers, 278
- Reversion/suppression, somatic, Duchenne muscular dystrophy, 950
- Rh segregation distortion, 1328
- Rhodopsin gene, codon 255/256 deletion, autosomal dominant retinitis pigmentosa, 876 (L)
- Rhodopsin retinitis pigmentosa, recombination, 590
- Ring chromosome formation, mechanisms, 15
- Risk, genetic, myotonic dystrophy, 1340 (L), 1341 (L)
- Risk calculation, X-linked diseases, 960
- Robertsonian translocations  
 Fluorescence in situ hybridization, 174  
 t(14q21q), breakpoint, 717
- Rod monochromacy, autosomal recessive, 690
- RP6, autosomal dominant retinitis pigmentosa, 634
- SAP-1 deficiency, sulfatide metabolism, 1252
- Satellite III DNA subfamily, chromosome 14, 706
- Screening  
 Carrier, cystic fibrosis, 439 (L)  
 Population, cystic fibrosis, 438 (L)
- Segregation analysis  
 Alzheimer disease, 645 (L), 646 (L)  
 Blood infection, malaria, 1308
- Segregation distortion, Rh, 1328
- Sex-chromosome pairing, pseudoautosomal region, 1162
- Sib-pair linkage analysis, apo B gene, 1038



- Skeletal muscle, mitochondrial encephalomyopathy, lactic acidosis, and strokelike episodes, 934
- Software, genetic linkage, 1267
- Somatic mosaicism, maternal age and Alzheimer disease, 1342 (L), 1343 (L)
- Spinal muscular atrophy  
Locus map, chromosome 5, 520  
Types II and III, proximal, chromosome 5q, 892
- Steroid sulfatase gene, point mutations, X-linked ichthyosis, 483
- Strokelike episodes, lactic acidosis, mitochondrial encephalomyopathy, 934
- Strokelike episodes, mitochondrial myopathy, encephalopathy, lactic acidosis, 655 (L)
- Substitution, Cys<sub>138</sub> to Arg, G<sub>M2</sub> activator protein, G<sub>M2</sub> gangliosidosis, 1046
- Sulfatide metabolism, SAP-1 deficiency, 1252
- T→G at 8993, Leigh disease, 852
- t(14q21q) Robertsonian translocations, breakpoint, 717
- T-cell X inactivation, maximum-likelihood analysis, 742
- Telomeric sequences, interstitial, 914
- Testis-determining region, sex phenotypes, 1008
- Thalassemia, β-  
Molecular characterization, 422  
- 32(C-A) mutant, 237 (L)
- Torsion dystonia, chromosome 9q34, 619
- Transforming growth factor alpha, genetic variation, 870 (L)
- Translocation(s)  
Reciprocal, chromosome painting, 700  
Robertsonian  
Breakpoint, 717  
Fluorescence in situ hybridization, 174  
X/autosome, sequence motifs, 725
- Transthyretin 122 (Val→Ile), PCR-PIRA, 195
- Transthyretin M<sup>119</sup>, and disease, 29
- Transthyretin mutation, amyloid cardiomyopathy, 1027
- Trisomy 15, uniparental disomy 15, 1348 (L)
- Trisomy 21  
Meiosis I and II nondisjunction, recombination, 1137 (L)  
Nondisjunction, 544
- Tritanopia, blue-sensitive opsin, 498
- tRNA<sup>Leu(UUR)</sup> mutation, mitochondrial, MELAS, 934
- Tubulopathy, proximal, mitochondrial genome duplication, 364
- Tumor genes, hereditary, carriers, 1350 (L)
- Tumorigenesis, medulloblastoma, chromosome 17p, 584
- Uniparental disomy 15, trisomy 15, 1348 (L)
- Uniparental isodisomy, maternal, chromosome 14, 690
- Variable number of tandem repeats, retinoblastoma gene, 371
- von Willebrand disease type IIB, germ-line mosaicism, 199
- von Willebrand factor, glycoprotein IIb-binding domain, germ-line mosaicism, 199
- Waardenburg syndrome type I, chromosome 2, 902
- Wilson, Allan C., 234 (Obit)
- X/autosome translocation, sequence motifs, 725
- X-linked cleft palate, *PGK1* and *DXYS1*, 1129
- X-linked combined immunodeficiency, carriers, 742
- X-linked diseases, germinal mosaicism and risk calculation, 960
- X-linked ichthyosis, steroid sulfatase gene point mutations, 483
- X-linked recessive modifier, Huntington disease age at onset, 536
- Xeroderma pigmentosum, phenotype, 677
- XK (McLeod locus), Xp21, 317
- XY275 *Low* allele, Y association, 1301
- Y-associated XY275 *Low* allele, 1301